Complete Summary

GUIDELINE TITLE

Screening for fetal chromosomal abnormalities.

BIBLIOGRAPHIC SOURCE(S)

American College of Obstetricians and Gynecologists (ACOG). Screening for fetal chromosomal abnormalities. Washington (DC): American College of Obstetricians and Gynecologists (ACOG); 2007 Jan. 11 p. (ACOG practice bulletin; no. 77). [43 references]

GUIDELINE STATUS

This is the current release of the guideline.

This guideline updates a previous version: American College of Obstetricians and Gynecologists (ACOG). Prenatal diagnosis of fetal chromosomal abnormalities. Washington (DC): American College of Obstetricians and Gynecologists (ACOG); 2001 May. 12 p.(ACOG practice bulletin; no. 27).

COMPLETE SUMMARY CONTENT

SCOPE

DISEASE/CONDITION(S)

Fetal chromosomal abnormalities including:

- Down syndrome
- Trisomy 18
- Trisomy 13
- Turner syndrome
- Neural tube defect

- Other significant fetal chromosome defects

GUIDELINE CATEGORY

Counseling
Diagnosis
Prevention
Risk Assessment
Screening

CLINICAL SPECIALTY

Medical Genetics
Obstetrics and Gynecology

INTENDED USERS

Physicians

GUIDELINE OBJECTIVE(S)

- To aid practitioners in making decisions about appropriate obstetric and gynecologic care
- To present and evaluate the best available evidence for the use of ultrasonographic and serum markers for selected aneuploidy screening in pregnancy
- To offer practical recommendations for implementing Down syndrome screening in practice

TARGET POPULATION

Pregnant women

INTERVENTIONS AND PRACTICES CONSIDERED

Screening

Laboratory Screening

1. Maternal serum alpha-fetoprotein (AFP)
2. Free or total beta-human chorionic gonadotropin (hCG)
3. Unconjugated estriol
4. Inhibin A
5. Pregnancy-associated plasma protein A (PAPP-A)

Imaging

1. Ultrasonography
2. Nuchal translucency (NT) measurement
3. Fetal echocardiogram (for Down syndrome-related anomalies)

Diagnostic Testing

1. Amniocentesis
2. Chorionic villus sampling (CVS)

Management

1. Patient counseling
2. Trimester-appropriate screening
3. Integrated (1^{st} and 2^{nd} trimester marker) screening
4. Stepwise sequential screening
5. Contingent sequential screening
6. Test interpretation
7. Management of multifetal gestations

MAJOR OUTCOMES CONSIDERED

- Risks and benefits of diagnostic procedures
- Predictive value of ultrasound markers of aneuploidy
- Predictive value of diagnostic tests for detection of fetal chromosomal abnormalities

METHODOLOGY

METHODS USED TO COLLECT/SELECT EVIDENCE

Hand-searches of Published Literature (Primary Sources)
Hand-searches of Published Literature (Secondary Sources)
Searches of Electronic Databases

DESCRIPTION OF METHODS USED TO COLLECT/SELECT THE EVIDENCE

The MEDLINE database, the Cochrane Library, and American College of Obstetricians and Gynecologists' (ACOG's) own internal resources and documents were used to conduct a literature search to locate relevant articles published between January 1985 and September 2006. The search was restricted to articles published in the English language. Priority was given to articles reporting results of original research, although review articles and commentaries also were consulted. Abstracts of research presented at symposia and scientific conferences were not considered adequate for inclusion in this document. Guidelines published by organizations or institutions such as the National Institutes of Health and the American College of Obstetricians and Gynecologists were reviewed, and additional studies were located by reviewing bibliographies of identified articles.

NUMBER OF SOURCE DOCUMENTS

Not stated

METHODS USED TO ASSESS THE QUALITY AND STRENGTH OF THE EVIDENCE

Weighting According to a Rating Scheme (Scheme Given)

RATING SCHEME FOR THE STRENGTH OF THE EVIDENCE

Studies were reviewed and evaluated for quality according to the method outlined by the U.S. Preventive Services Task Force:

I Evidence obtained from at least one properly designed randomized controlled trial.

II-1 Evidence obtained from well-designed controlled trials without randomization.

II-2 Evidence obtained from well-designed cohort or case–control analytic studies, preferably from more than one center or research group.

II-3 Evidence obtained from multiple time series with or without the intervention. Dramatic results in uncontrolled experiments also could be regarded as this type of evidence.

III Opinions of respected authorities, based on clinical experience, descriptive studies, or reports of expert committees.

METHODS USED TO ANALYZE THE EVIDENCE

Review of Published Meta-Analyses
Systematic Review

DESCRIPTION OF THE METHODS USED TO ANALYZE THE EVIDENCE

Not stated

METHODS USED TO FORMULATE THE RECOMMENDATIONS

Expert Consensus

DESCRIPTION OF METHODS USED TO FORMULATE THE RECOMMENDATIONS

Analysis of available evidence was given priority in formulating recommendations. When reliable research was not available, expert opinions from obstetrician-gynecologists were used. See also the "Rating Scheme for the Strength of Recommendations" field regarding Grade C recommendations.

RATING SCHEME FOR THE STRENGTH OF THE RECOMMENDATIONS

Based on the highest level of evidence found in the data, recommendations are provided and graded according to the following categories:

Level A — Recommendations are based on good and consistent scientific evidence.

Level B — Recommendations are based on limited or inconsistent scientific evidence.

Level C — Recommendations are based primarily on consensus and expert opinion.

COST ANALYSIS

A formal cost analysis was not performed and published cost analyses were not reviewed.

METHOD OF GUIDELINE VALIDATION

Internal Peer Review

DESCRIPTION OF METHOD OF GUIDELINE VALIDATION

Practice Bulletins are validated by two internal clinical review panels composed of practicing obstetrician-gynecologists generalists and subspecialists. The final guidelines are also reviewed and approved by the American College of Obstetricians and Gynecologists (ACOG) Executive Board.

RECOMMENDATIONS

MAJOR RECOMMENDATIONS

The grades of evidence (I-III) and levels of recommendations (A-C) are defined at the end of "Major Recommendations" field.

The following recommendation is based on good and consistent scientific evidence (Level A):

- First-trimester screening using both nuchal translucency measurement and biochemical markers is an effective screening test for Down syndrome in the general population. At the same false-positive rates, this screening strategy results in a higher Down syndrome detection rate than does the second-trimester maternal serum triple screen and is comparable to the quadruple screen.
- Measurement of nuchal translucency alone is less effective for first-trimester screening than is the combined test (nuchal translucency measurement and biochemical markers).
- Women found to have increased risk of aneuploidy with first-trimester screening should be offered genetic counseling and the option of chorionic villus sampling (CVS) or second-trimester amniocentesis.
- Specific training, standardization, use of appropriate ultrasound equipment, and ongoing quality assessment are important to achieve optimal nuchal

translucency measurement for Down syndrome risk assessment, and this procedure should be limited to centers and individuals meeting these criteria.
- Neural tube defect screening should be offered in the second trimester to women who elect only first-trimester screening for aneuploidy.

The following recommendations are based on limited or inconsistent scientific evidence (Level B):

- Screening and invasive diagnostic testing for aneuploidy should be available to all women who present for prenatal care before 20 weeks of gestation regardless of maternal age. Women should be counseled regarding the differences between screening and invasive diagnostic testing.
- Integrated first- and second-trimester screening is more sensitive with lower false-positive rates than first-trimester screening alone.
- Serum integrated screening is a useful option in pregnancies where nuchal translucency measurement is not available or cannot be obtained.
- An abnormal finding on second-trimester ultrasound examination identifying a major congenital anomaly significantly increases the risk of aneuploidy and warrants further counseling and the offer of a diagnostic procedure.
- Patients who have a fetal nuchal translucency measurement of 3.5 mm or higher in the first trimester, despite a negative aneuploidy screen, or normal fetal chromosomes, should be offered a targeted ultrasound examination, fetal echocardiogram, or both.
- Down syndrome risk assessment in multiple gestation using first- or second-trimester serum analytes is less accurate than in singleton pregnancies.
- First-trimester nuchal translucency screening for Down syndrome is feasible in twin or triplet gestation but has lower sensitivity than first-trimester screening in singleton pregnancies.

The following recommendations are based primarily on consensus and expert opinion (Level C):

- After first-trimester screening, subsequent second-trimester Down syndrome screening is not indicated unless it is being performed as a component of the integrated test, stepwise sequential, or contingent sequential test.
- Subtle second-trimester ultrasonographic markers should be interpreted in the context of a patient's age, history, and serum screening results.

Definitions:

Grades of Evidence

I Evidence obtained from at least one properly designed randomized controlled trial.

II-1 Evidence obtained from well-designed controlled trials without randomization.

II-2 Evidence obtained from well-designed cohort or case–control analytic studies, preferably from more than one center or research group.

II-3 Evidence obtained from multiple time series with or without the intervention. Dramatic results in uncontrolled experiments also could be regarded as this type of evidence.

III Opinions of respected authorities, based on clinical experience, descriptive studies, or reports of expert committees.

Levels of Recommendation

Level A — Recommendations are based on good and consistent scientific evidence.

Level B — Recommendations are based on limited or inconsistent scientific evidence.

Level C — Recommendations are based primarily on consensus and expert opinion.

CLINICAL ALGORITHM(S)

None provided

EVIDENCE SUPPORTING THE RECOMMENDATIONS

TYPE OF EVIDENCE SUPPORTING THE RECOMMENDATIONS

The type of supporting evidence is identified and graded for each recommendation (see "Major Recommendations").

BENEFITS/HARMS OF IMPLEMENTING THE GUIDELINE RECOMMENDATIONS

POTENTIAL BENEFITS

Appropriate prenatal diagnosis of fetal chromosomal abnormalities

POTENTIAL HARMS

- Loss of fetus from invasive diagnostic testing
- False positive chromosomal abnormality results

QUALIFYING STATEMENTS

QUALIFYING STATEMENTS

These guidelines should not be construed as dictating an exclusive course of treatment or procedure. Variations in practice may be warranted based on the needs of the individual patient, resources, and limitations unique to the institution or type of practice.

IMPLEMENTATION OF THE GUIDELINE

DESCRIPTION OF IMPLEMENTATION STRATEGY

An implementation strategy was not provided.

IMPLEMENTATION TOOLS

Audit Criteria/Indicators
Patient Resources

For information about availability, see the "Availability of Companion Documents" and "Patient Resources" fields below.

INSTITUTE OF MEDICINE (IOM) NATIONAL HEALTHCARE QUALITY REPORT CATEGORIES

IOM CARE NEED

Staying Healthy

IOM DOMAIN

Effectiveness
Patient-centeredness
Timeliness

IDENTIFYING INFORMATION AND AVAILABILITY

BIBLIOGRAPHIC SOURCE(S)

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ADAPTATION

Not applicable: The guideline was not adapted from another source.

DATE RELEASED

2001 May (revised 2007 Jan)

GUIDELINE DEVELOPER(S)

American College of Obstetricians and Gynecologists - Medical Specialty Society

SOURCE(S) OF FUNDING

American College of Obstetricians and Gynecologists (ACOG)

GUIDELINE COMMITTEE

American College of Obstetricians and Gynecologists (ACOG) Committee on Practice Bulletins-Obstetrics

American College of Obstetricians and Gynecologists (ACOG) Committee on Genetics

Society for Maternal-Fetal Medicine Publications Committee

COMPOSITION OF GROUP THAT AUTHORED THE GUIDELINE

Not stated

FINANCIAL DISCLOSURES/CONFLICTS OF INTEREST

Not stated

GUIDELINE STATUS

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GUIDELINE AVAILABILITY

Electronic copies: None available

Print copies: Available for purchase from the American College of Obstetricians and Gynecologists (ACOG) Distribution Center, PO Box 4500, Kearneysville, WV 25430-4500; telephone, 800-762-2264, ext. 192; e-mail: sales@acog.org. The ACOG Bookstore is available online at the ACOG Web site.

AVAILABILITY OF COMPANION DOCUMENTS

Proposed performance measures are included in the original guideline document.

PATIENT RESOURCES

The following is available:

- Genetic disorders. Atlanta (GA): American College of Obstetricians and Gynecologists (ACOG); 2005.

Electronic copies: Available from the American College of Obstetricians and Gynecologists (ACOG) Web site.

Print copies: Available for purchase from the American College of Obstetricians and Gynecologists (ACOG) Distribution Center, PO Box 4500, Kearneysville, WV 25430-4500; telephone, 800-762-2264, ext. 192; e-mail: sales@acog.org. The ACOG Bookstore is available online at the ACOG Web site.

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NGC STATUS

This NGC summary was completed by ECRI on September 22, 2004. The information was verified by the guideline developer on December 9, 2004. This NGC summary was updated by ECRI Institute on October 8, 2007. The updated information was verified by the guideline developer on December 3, 2007.

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